GATA1 gene

GATA binding protein 1

Normal Function

The *GATA1* gene provides instructions for making a protein that attaches (binds) to specific regions of DNA and helps control the activity of many other genes. On the basis of this action, the GATA1 protein is known as a transcription factor. The GATA1 protein is involved in the specialization (differentiation) of immature blood cells. To function properly, these immature cells must differentiate into specific types of mature blood cells. By binding to DNA and interacting with other proteins, the GATA1 protein regulates the growth and division (proliferation) of immature red blood cells and platelet-precursor cells (megakaryocytes) to facilitate their differentiation. Red blood cells help carry oxygen to various tissues throughout the body and platelets aid in blood clotting. The GATA1 protein is also important for the maturation of several types of white blood cells that help fight infection, including eosinophils, mast cells, and dendritic cells.

Two versions of the GATA1 protein are produced from the *GATA1* gene: a regular length protein and a shorter version called GATA1s. The GATA1s protein lacks a specific region called the transactivation domain. Although the specific function of this region is unclear, researchers believe that it interacts with other proteins to modify GATA1 protein function.

Health Conditions Related to Genetic Changes

dyserythropoietic anemia and thrombocytopenia

At least eight different mutations in the *GATA1* gene have been found to cause dyserythropoietic anemia and thrombocytopenia. Most of these mutations change a single protein building block (amino acid) in the GATA1 protein. *GATA1* gene mutations disrupt the protein's ability to bind with DNA or interact with other proteins. This impairment in the GATA1 protein's normal function leads to increased proliferation,decreased differentiation, and premature death of immature blood cells. Immature blood cells cannot perform the functions of specialized, mature blood cells. A lack of differentiation causes a shortage of red blood cells (anemia) and platelets involved in blood clotting (thrombocytopenia), which are characteristic features of dyserythropoietic anemia and thrombocytopenia.

cancers

Some gene mutations can be acquired during a person's lifetime and are present only in certain cells. These mutations are called somatic mutations, and they are not

inherited. Somatic mutations in the *GATA1* gene increase the risk of developing a disease of blood-forming cells called transient abnormal myelopoiesis (TAM). These mutations usually occur during fetal development, and the increased risk only applies to people who are born with an extra copy of chromosome 21 in each of their cells, a condition known as trisomy 21 or Down syndrome.

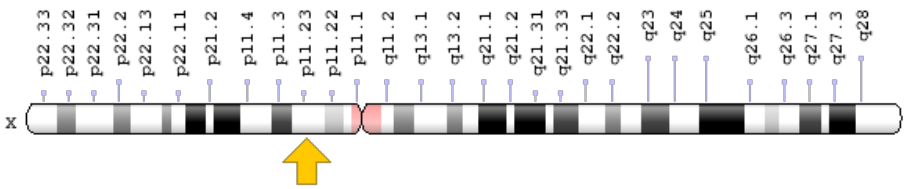
Approximately 10 percent of people with Down syndrome develop TAM, usually at birth or soon after. TAM is characterized by the accumulation of immature megakaryocyte precursor cells in the blood, liver, and bone marrow. In most cases, TAM causes no signs or symptoms and disappears within three to four months. However, approximately 20 percent of infants with TAM will have serious complications such as an enlarged liver (hepatomegaly), a buildup of scar tissue in the liver (hepatic fibrosis), excess fluid accumulation in the body before birth (hydrops fetalis), and heart failure.

It is estimated that 20 to 30 percent of children with TAM will later develop a cancer of the blood-forming cells called acute megakaryoblastic leukemia (AMKL). The somatic *GATA1* gene mutations found in individuals with TAM or AMKL prevent the production of the normal length GATA1 protein and only allow production of the shorter version, GATA1s. It is unclear why somatic *GATA1* gene mutations increase the risk of developing these bone marrow disorders in people with Down syndrome.

Chromosomal Location

Cytogenetic Location: Xp11.23, which is the short (p) arm of the X chromosome at position 11.23

Molecular Location: base pairs 48,786,540 to 48,794,311 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ERYF1
- erythroid transcription factor
- erythroid transcription factor 1

- GATA-1
- GATA-binding factor 1
- GATA binding protein 1 (globin transcription factor 1)
- GATA1_HUMAN
- GF-1
- GF1
- globin transcription factor 1
- transcription factor GATA1

Additional Information & Resources

GeneReviews

- GATA1-Related X-Linked Cytopenia
  https://www.ncbi.nlm.nih.gov/books/NBK1364

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28GATA1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- DOWN SYNDROME
  http://omim.org/entry/190685
- GATA-BINDING PROTEIN 1
  http://omim.org/entry/305371

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GATA1ID40689chXp11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GATA1%5Bgene%5D
- HGNC Gene Family: GATA zinc finger domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/82
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4170

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2623
- UniProt
  http://www.uniprot.org/uniprot/P15976

Sources for This Summary

- Cabelof DC, Patel HV, Chen Q, van Remmen H, Matherly LH, Ge Y, Taub JW. Mutational spectrum at GATA1 provides insights into mutagenesis and leukemogenesis in Down syndrome. Blood. 2009 Sep 24;114(13):2753-63. doi: 10.1182/blood-2008-11-190330. Epub 2009 Jul 24.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19633202
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2756130/
- Crispino JD. GATA1 in normal and malignant hematopoiesis. Semin Cell Dev Biol. 2005 Feb;16(1): 137-47. Epub 2004 Dec 13. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15659348
- Ferreira R, Ohneda K, Yamamoto M, Philipsen S. GATA1 function, a paradigm for transcription factors in hematopoiesis. Mol Cell Biol. 2005 Feb;25(4):1215-27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15684376
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC548021/
- OMIM: GATA-BINDING PROTEIN 1
  http://omim.org/entry/305371
- Kobayashi M, Yamamoto M. Regulation of GATA1 gene expression. J Biochem. 2007 Jul;142(1): 1-10. Epub 2007 Jun 13. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17567656
- Shimizu R, Engel JD, Yamamoto M. GATA1-related leukaemias. Nat Rev Cancer. 2008 Apr;8(4): 279-87. doi: 10.1038/nrc2348. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18354416
- Splendore A, Magalhães IQ, Pombo-de-Oliveira MS. GATA1 mutations in myeloproliferative disorders: nomenclature standardization and review of the literature. Hum Mutat. 2005 Oct;26(4): 390-2. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16134163
- Vardiman JW, Thiele J, Arber DA, Brunning RD, Borowitz MJ, Porwit A, Harris NL, Le Beau MM, Hellström-Lindberg E, Tefferi A, Bloomfield CD. The 2008 revision of the World Health Organization (WHO) classification of myeloid neoplasms and acute leukemia: rationale and important changes. Blood. 2009 Jul 30;114(5):937-51. doi: 10.1182/blood-2009-03-209262. Epub 2009 Apr 8. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19357394

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/GATA1

Reviewed: July 2010
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services